Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Solid Tumour Section

Short Communication

t(1;22)(q23;q12) in myoepithelioma

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Clinics and pathology

Disease

Myoepitheliomas are tumours which can occur in the salivary glands, in soft tissues and other organs, with a variable but generally low to intermediate aggressiveness (myoepithelial carcinoma for the most adverse histology).

Epidemiology

One case to date, a 59-year-old female patient with a 10 years long history. The patient was well 7 months after treatment of a myoepithelioma with an uncertain degree of malignancy (Brandal et al., 2008).

Cytogenetics

Cytogenetics Morphological

The t(1;22)(q23;q12) was the sole anomaly.

Genes involved and proteins

PBX1

Location: 1q23

Protein

Homeobox protein (homeodomain in amino acids 233-295). Binds the sequence 5'-ATCAATCAA-3'. Transcription factor.

EWSR1

Location: 22q12

Protein

From N-term to C-term: a transactivation domain (TAD) containing multiple degenerate hexapeptide repeats, 3 arginine/glycine rich domains (RGG regions), a RNA recognition motif, and a RanBP2 type Zinc finger. Role in transcriptional regulation for specific genes and in mRNA splicing.

Result of the chromosomal anomaly

Hybrid Gene

Description

5' EWSR1 - 3' FLI1. EWSR1 exon 7 is fused in frame to PBX1 exon 5.

Fusion Protein

Description

Fusion of the N terminal transactivation domain of EWSR1 to the homeobox (DNA binding domain) of PBX1.

References

Brandal P, Panagopoulos I, Bjerkehagen B, Gorunova L, Skjeldal S, Micci F, Heim S. Detection of a t(1;22)(q23;q12) translocation leading to an EWSR1-PBX1 fusion gene in a myoepithelioma. Genes Chromosomes Cancer. 2008 Jul;47(7):558-64

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